What's new

SEQUENCE Pilot CE

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developed by

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1 All modules

1.1 License

The message you get in case your program is unlicensed changed. You are now asked which module you want to test. Moreover the path of the `lis.ini` file is listed.

2 Modules SeqPatient, SeqNext and SeqC

2.1 Gene Admin

In case an isoform is selected in *Gene Admin*, the isoform is pre-selected when the dialogue *Change Gene* is opened (*[Extras] > change gene*).

2.2 Operation Mutation [master file]

- New button *[Organ/Phenotype …]*: Here organs and phenotypes can be defined. These can be added to mutations listed in the mutation database (dialogue *Mutation*, see below).
- Dialogue *Mutation*: The dialogue has been worked over:
  - *Effect* was renamed into *Mut Effect.*
  - *p. mut Effect:* Here a percent value of a mutation effect can be selected.
  - *Ethnicity:* It's possible to choose an ethnicity in the drop down list. Only ethnicities that are added in the *Orders* table of this dialogue are available (left-click into the column *order*)!
  - *p. mut Causality:* Here a percent value of a mutation causality can be selected.
  - *Organ/Phenotype table*: Defined organs/phenotypes can be added to or removed from mutations listed in the mutation database.
- New table column *Zygosity* in the *Orders* table. Entry is *homo* for a homozygous mutation and *het* for a heterozygous mutation.

3 Modules SeqNext and SeqHLA454

3.1 Operation Run/Automatic start of a run

A run can be started automatically after files are analysed on your sequencer. Therefore all parameters needed for the operation *Run* have to be defined in a `txt`-file. The `txt`-file has to be saved in the directory `bin\Autorun\SeqNext` (for SeqNext) or `bin\Autorun\SeqNextHLA` (for SeqHLA454) of your **SEQUENCE Pilot** installation. For further information about the `txt`-file see **User Manual SeqNext / chapter 8.3.4.5 Automatic start of a Run** or contact our support team.

3.2 Operation Joining

Result files can be joined to medical validated or archived orders. Therefore enter `JoinRFValidatedOrder=YES` in the section `[SeqNext]` or `[SeqNextHLA]` respectively.
Archived orders are then reactivated automatically in case further data is imported for this patient. The new result file is joined to the order and the state *MV* is removed.
If you want to create a new patient order in case the order is medically validated, enter additionally `CreateOrderByRun=YES` or `CreateOrderByProject=YES` (in case you use projects) in the section `[SeqNext]` or `[SeqNextHLA]` respectively.

3.3 Operation Archiving

During archiving an empty file is saved in your **SEQUENCE Pilot** installation, which shows that an order was archived. Therefore for all archived orders the file `archive.txt` is present in the corresponding folder in the directory `SeqNResults` of your installation.

4 Modules SeqPatient and SeqHLA

4.1 Operation SeqPrimer [master file]

The automatic naming of **forward** and **reverse** direction in the *SeqPrimer Name* (by default **F** (forward) and **R** (reverse)) can be changed in the `lis.ini` file, located in the `bin`-directory of your installation:

Therefore enter the desired character in the `lis.ini`-section `[SeqPilot]` behind the following entries:

```
SeqPrimerForwardDescription=
SeqPrimerReverseDescription=
```

e.g.: for lower case **f** for forward primer and lower case **r** for reverse primer enter:

```
SeqPrimerForwardDescription=f
SeqPrimerReverseDescription=r
```

5 Module SeqNext

5.1 Operations Poollist and Pool

Patient pools can be analysed using the operations *Poollist* and *Pool*. See **User Manual SeqNext / chapter 10 Patient Pool analysis** for detailed information.

5.2 Operation PCR Groups [master file]

New field *Control* is available. This is only relevant for CNV analysis. Here the controls that should be used for an analysis (date or default) can be selected.

5.3 Operation Run

- The sequencing platform used, has to be selected in the dialogue *Settings*. The following platforms are available: *454, Ion Torrent, MiSeq, Solexa, SOLID*.
- Two barcodes to identify fragments of **one** patient can be entered into the field *Barcode* in the following way:
  - *barcode1,barcode2*: fragments are labelled with barcode 1 **or** barcode 2
  - *barcode1,-barcode 2*: fragments start with barcode 1 **and ends** with barcode 2.
- New table *Organ/Phenotype* to add an organ/phenotype to be analysed. Only organs/phenotypes that are pre-defined in the operation *Mutation [master file]* can be added.

5.4 Operation Joining

- New item *Org./P.* to search for orders with defined organs/phenotypes in the dialogue part *Select Orders*.

- Context menu item *settings...* is not available any more in the *Lower Table* (only in the *Upper table*).
- Context menu item *settings...* in the *Upper Table*: The dialogue *File settings* includes the *Organ/Phenotype table*. Organs/Phenotypes can be added or removed by pressing the corresponding button. Only organs/phenotypes that are pre-defined in the operation *Mutation [master file]* can be added.

5.5 Operation Orderlist/File line

In case several files are loaded in one *Run* the file names are listed in the *file line* separated by ,.For one file loaded in one *Run* the path of the file is listed.

5.6 Operation Joining and operation Worklist, column File

In case several files are loaded in one *Run* the file names are listed in this column separated by ,. For one file loaded in one *Run* the path of the file is listed.

5.7 Operation Worklist/Select Orders

New item *Org./P.* to search for orders with defined organs/phenotypes.

5.8 Operation Sequence/Files

Several files loaded in one *Run* are listed in one line. The number of files (*1 file* or *2 files*...) is shown in the column *File*. Moreover in case only **one** file is loaded the file name is listed.

The context menu item *show* > *info* has been worked over. The information shows the path of all files loaded in one *Run*.

New context menu item *show* > *organ/phenotype...* to show a list of organs/phenotypes added to the file.

5.9 Operation Sequence/Electropherogram

New context menu item *copy* in the gene sequence and the result file sequences:

- *seq. part:* copies the sequence shown on the screen.
- *seq. complete*: copies the sequence of the complete *Gene/Chromosome*.

5.10 Operation Sequence/CNV window

In the control settings (button *control settings*), tab *Selections*, archived controls are not available any longer.

5.11 Operation Archiving/Select Orders

New item *Org./P.* to search for orders with defined organs/phenotypes.

6 Module SeqPatient

6.1 Operation Sequence/dialogue Edit Bases

Context menu for a selected position in the gene line:

2nd allele/Delete bases... and *2nd allele/Insert bases...*: The maximum number of bases that can be inserted/deleted is *1000*.

7 Module SeqHLA

7.1 HLA SBT kits from R.O.S.E.

For **HLA SBT Kits** from **R.O.S.E.** pre-defined amplification modules and sequencing primers are available to download on our homepage http://www.jsi-medisys.de/rosekits.html. Those can be added to your installation, following the instructions after executing the downloaded file.

8 Module Talkmaster

8.1 Export of data

MLPA data, SeqNext data and SeqHLA 454 data can be exported.